CLAIMS

1. An isolated nucleic acid encoding UDP-N-Acetylglucosamine: Galactose-β1,3-N-Acetylgalactosamine-α-R β1-6 N-Acetylglucosaminyltransferase (C2GnT3) or a fragment thereof.

2. The isolated nucleic acid of claim 1, wherein said nucleic acid is DNA.

3. The isolated nucleic acid of claim 2, wherein said DNA is cDNA.

4. The isolated nucleic acid of claim 2, wherein said DNA is genomic DNA.

5. The isolated nucleic acid of claim 1, wherein said isolated nucleic acid comprises the nucleotide sequence of SEQ ID NO: 1, or a sequence-conservative or function-conservative variant thereof.

6. A nucleic acid which hybridizes under conditions of high stringency with a nucleic acid comprising the nucleotide sequence of SEQ ID NO: 1.

7. An expression vector comprising a nucleotide sequence encoding C2GnT3 or a fragment thereof.

8. The vector of claim 7, wherein said nucleotide sequence comprises the nucleotide sequence of SEQ ID NO: 1, or a sequence-conservative or function-conservative variant thereof.

9. The vector of claim 7, wherein said sequence encoding C2GnT3 is operably linked to a transcriptional regulatory element.

10. A cell comprising the vector of claim 7.

11. The cell of claim 10, wherein said cell is stably transfected with said vector.

12. The cell of claim 10, wherein said cell produces enzymatically active C2GnT3.

13. The cell of claim 10, wherein said cell is selected from the group consisting of bacterial, yeast, insect, avian, and mammalian cells.

14. The cell in claim 10, wherein said cell is Sf9.

15. The cell in claim 10, wherein said cell is CHO.

16. A method for producing C2GnT3 polypeptides, which method comprises:

(i) introducing into a host cell an isolated DNA molecule encoding human C2GnT3, or a DNA construct comprising a DNA sequence encoding C2GnT3;

(ii) growing the host cell under conditions suitable for human C2GnT3 expression; and

(iii) isolating C2GnT3 produced by the host cell.

17. The method of claim 16, wherein said enzymatically active C2GnT3 is selected from the group consisting of:

(i) a polypeptide comprising the amino acid sequence of SEQ ID NO: 2;

(ii) a polypeptide comprising amino acid residues 39-453 of SEQ ID NO: 2;

(iii) a polypeptide comprising amino acids residues 39-453 of SEQ ID NO: 2 fused to a second sequence, wherein said second sequence comprises an affinity ligand or a reactive group; and

(iv) a function-conservative variant of any of the polypeptides in (i) to (iii).

18. A method of detecting whether at least one agent is an inhibitor or stimulator of C2GnT3 enzymatic activity in a cell-free or cell-based assay, which method comprises:

(i) contacting a C2GnT3 polypeptide in claim 17 with said at least one agent under assay conditions suitable for the detection of said enzymatic activity; and

(ii) measuring whether said enzymatic activity is inhibited or stimulated by said at least one agent,

wherein said at least one agent is selected from compounds, compositions, antibodies or antibody fragments, antisense sequences and ribozyme nucleotide sequences for C2GnT3 polypeptide.

19. A method of detecting whether at least one agent is an inhibitor or stimulator of C2GnT3 enzymatic activity in a cell-free or cell-based assay, which method comprises:

(i) contacting a cell that recombinantly expresses C2GnT3 polypeptide according to claims 10 with said at least one agent under assay conditions suitable for the detection of said enzymatic activity; and

(iii) measuring whether said enzymatic activity is inhibited or stimulated by said at least one agent,

wherein said at least one agent is selected from compounds, compositions, antibodies or antibody fragments, antisense sequences and ribozyme nucleotide sequences for C2GnT3 polypeptide.

20. The method of claim 18, wherein said at least one agent is a member of a combinatorial chemical library.

21. The method of claim 19, wherein said at least one agent is a member of a combinatorial chemical library.

22. The method of claim 18, wherein said at least one agent is generated by methods of C2GnT3 polypeptide structure-based design.

23. The method of claim 19, wherein said at least one agent is generated by methods of C2GnT3 polypeptide structure-based design.

24. A method for identifying DNA sequence variations in the C2GnT3 gene, comprising the steps of:

(i) isolating DNA from a patient;

(ii) amplifying at least one C2GnT3 genomic region from said DNA by PCR; and

(iii) analyzing said amplified C2GnT3 genomic region by DNA sequencing, single-strand conformational polymorphism (SSCP) or mismatch mutation, to detect whether a sequence variation vis-à-vis SEQ ID NO: 1 exists.

25. An isolated C2GnT3 polypeptide comprising the amino acid sequence of SEQ ID NO: 2.

26. An isolated polypeptide having at least 45% amino acid sequence identity to the amino acid sequence of SEQ ID NO: 2.

27. A polypeptide produced by the method of claim 16.

28. An antibody specifically recognizing a polypeptide of any of claims 25 to 27.

29. A probe comprising a sequence encoding a polypeptide of any of claims 25 to 27, or a part of a polypeptide of any of claims 25 to 27.

30. A method of diagnosing or monitoring conditions mediated by a C2GnT3 polypeptide comprising determining the presence of the nucleic acid molecule of claim 1 in a biological sample.

31. The method of claim 30, wherein the condition is a thymus-related disorder.

32. A method of diagnosing or monitoring conditions mediated by a C2GnT3 polypeptide comprising determining the presence of the nucleic acid molecule of claim 6 in a biological sample.

33. The method of claim 32 wherein the condition is a thymus-related disorder.

34. A method of diagnosing or monitoring conditions mediated by a C2GnT3 polypeptide comprising determining the presence of the nucleic acid molecule of claim 7 in a biological sample.

35. The method of claim 34 wherein the condition is a thymus-related disorder.

36. A method of diagnosing or monitoring conditions mediated by a C2GnT3 polypeptide comprising determining the presence in a biological sample of a nucleic acid molecule in claim 25.

37. The method of claim 36, wherein the condition is a thymus-related disorder.

38. A method of diagnosing or monitoring conditions mediated by a C2GnT3 polypeptide comprising determining the presence in a biological sample of a nucleic acid molecule in claim 26.

39. The method of claim 38, wherein the condition is a thymus-related disorder.

40. A method of diagnosing or monitoring conditions mediated by a C2GnT3 polypeptide comprising determining the presence in a biological sample of a nucleic acid molecule in claim 27.

41. The method of claim 40, wherein the condition is a thymus-related disorder.

42. A method for identifying a substance which associates with a polypeptide in any one of claims 22 to 24 comprising:

(i) reacting the polypeptide with at least one substance which potentially can associate with the polypeptide, under conditions which permit the association between the substance and polypeptide; and

(ii) detecting polypeptide associated with the substance, wherein detection of associated polypeptide and substance indicates that the substance associated with the polypeptide.

43. A method for evaluation a compound for its ability to modulate the biological activity of a polypeptide in any one of claims 25 to 27 comprising providing a known concentration of the polypeptide with a substance which associates with the polypeptide, and a test compound under conditions which permit the formation of complexes between the substance and polypeptide, and detecting said complexes.

44. A method for detecting a nucleic acid molecule encoding a C2GnT3 polypeptide in a biological sample comprising the steps of:

(i) hybridizing a nucleic acid molecule of claim 1 to the nucleic acid in the biological sample; and

(ii) detecting whether a hybridization complex has been formed,

wherein the presence of a hybridization complex correlates with the presence of a nucleic acid molecule encoding the polypeptide in the biological sample.

45. The method in claim 44 wherein nucleic acids of the biological sample are amplified by the polymerase chain reaction prior to the hybridizing step.

46. A method for detecting a nucleic acid molecule encoding a C2GnT3 polypeptide in a biological sample comprising the steps of:

(i) hybridizing the nucleic acid molecule of claim 6 to the nucleic acid in the biological sample; and

(ii) detecting whether a hybridization complex has been formed,

wherein the presence of a hybridization complex correlates with the presence of a nucleic acid molecule encoding the polypeptide in the biological sample.

47. The method in claim 46 wherein nucleic acids of the biological sample are amplified by the polymerase chain reaction prior to the hybridizing step.

48. A method for detecting a nucleic acid molecule encoding a C2GnT3 polypeptide in a biological sample comprising the steps of:

(i) hybridizing the nucleic acid molecule of claim 7 to the nucleic acid in the biological sample; and

(ii) detecting whether a hybridization complex has been formed,

wherein the presence of a hybridization complex correlates with the presence of a nucleic acid molecule encoding the polypeptide in the biological sample.

49. The method in claim 48 wherein nucleic acids of the biological sample are amplified by the polymerase chain reaction prior to the hybridizing step.

50. A method for treating a condition mediated by a C2GnT3 polypeptide in any one of claims 25 to 27, comprising administering an effective amount of the antibody in claim 28, or a substance or compound identified using a method in any of claims 42 and 43.

51. A method as claimed in claim 50 wherein the condition is a thymus-related disorder.

52. A composition comprising one or more of a nucleic acid molecule in any of claims 1, 6, and 7, or the polypeptide in any of claims 25 to 27, or a substance or compound identified using a method in any of claims 18, 19, 42 and 43, and a pharmaceutically acceptable carrier, excipient or diluent.

53. Use of at least one nucleic acid molecule in any of claims 1, 6, and 7, or the polypeptide in any of claims 25 to 27, or a substance or compound identified using a method as claimed in any one of claims 18, 19, 42 and 43 in the preparation of a pharmaceutical composition for treating a condition mediated by a C2GnT3 polypeptide.

54. A gene-based therapy directed at the thymus comprising a polynucleotide comprising all or a portion of a regulatory sequence of SEQ ID NO: 1.

55. A method for preparing an oligosaccharide comprising contacting a reaction mixture comprising an activated GlcNAc, and an acceptor in the presence of an enzymatically active polypeptide as claimed in any one of claims 25 to 27.

56. An isolated nucleic acid comprising a nucleic acid segment free of internal non-coding sequences, said segment encoding UDP-N-Acetylglucosamine: Galactose-β1,3-N-Acetyl-galactosamine-α-R β1-6 N-Acetylglucosaminyl-transferase (C2GnT3) or a fragment thereof.

57. A method of detecting whether at least one agent is an inhibitor or stimulator of C2GnT3 enzymatic activity, which method comprises:

(i) contacting said at least one agent, under assay conditions suitable for the detection of said enzymatic activity, with a C2GnT3 polypeptide selected from the group consisting of a polypeptide comprising the amino acid sequence of SEQ ID NO: 2; a polypeptide comprising amino acid residues 39-453 of SEQ ID NO: 2; a polypeptide comprising amino acids residues 39-453 of SEQ ID NO: 2 fused to a

second sequence, wherein said second sequence comprises an affinity ligand or a reactive group; and a function-conservative variants of any of the foregoing polypeptides; and

(ii) measuring whether said enzymatic activity is inhibited or stimulated compared to a control value by said at least one agent.

58. The method of claim 57, wherein said at least one agent is selected from the group consisting of compounds, compositions, antibodies, antibody fragments, antisense sequences, and ribozyme nucleotide sequences for C2GnT3 polypeptide.

59. The method of claim 57, wherein said contacting takes place in a cell-free or a cell-based assay system.

60. An isolated polypeptide having at least 95% amino acid sequence identity to the amino acid sequence of SEQ ID NO: 2, and C2GnT3 enzymatic activity.

61. An antibody specifically recognizing the polypeptide in claim 60.

62. An isolated UDP-N-Acetylglucosamine: Galactose-β1,3-N-Acetylgalactosamine-α-R β1,6 N-Acetylglucosaminyltransferase (C2GnT) polypeptide comprising the amino acid sequence of residues 39-453 of SEQ ID NO: 2.

63. The isolated C2GnT polypeptide of claim 62, comprising the amino acid sequence of SEQ ID NO:2.

64. The isolated C2GnT polypeptide of claim 62, having the amino acid sequence of SEQ ID NO:2.

65. An isolated C2GnT polypeptide having at least 45% amino acid sequence identity to the amino acid sequence of SEQ ID NO: 2.

66. The isolated C2GnT polypeptide of claim 65, wherein said amino acid sequence identity is at least 60%.

67. The isolated C2GnT polypeptide of claim 65, wherein said amino acid sequence identity is at least 95%.

68. An isolated C2GnT polypeptide having at least 45% amino acid sequence identity to a human C2GnT enzyme which is expressed *in vivo* at a higher level in thymus tissue than in tracheal and thyroid tissue.

69. The isolated C2GnT polypeptide of claim 68, wherein said amino acid sequence identity is at least 60%.

70. The isolated C2GnT polypeptide of claim 68, wherein said amino acid sequence identity is at least 95%.

71. An isolated polypeptide having at least 95% amino acid sequence identity to SEQ ID NO:2 and C2GnT enzymatic activity.

72. A C2GnT polypeptide produced by a method comprising:

(i) introducing into a host cell an isolated DNA molecule encoding a human C2GnT polypeptide, or a DNA construct comprising a DNA sequence encoding a C2GnT polypeptide;

(ii) growing the host cell under conditions suitable for human C2GnT expression; and

(iii) isolating C2GnT polypeptide produced by the host cell,

wherein said C2GnT polypeptide is at least 45% identical to SEQ ID NO:2.

73. The C2GnT polypeptide of claim 72, wherein said C2GnT polypeptide is at least 60% identical to SEQ ID NO:2.

74. The C2GnT polypeptide of claim 72, wherein said C2GnT polypeptide is at least 95% identical to SEQ ID NO:2.

75. A method for preparing an oligosaccharide comprising contacting a compound comprising an activated GlcNAc, an acceptor, and the C2GnT polypeptide of any of claims 62, 65, 68, 71, and 72.

76. A method for preparing an oligosaccharide comprising contacting a compound comprising an activated GlcNAc, an acceptor, and a C2GnT polypeptide comprising the amino acid sequence of residues 39-453 of SEQ ID NO: 2.

77. The method of claim 76, wherein the C2GnT polypeptide comprises the amino acid sequence of SEQ ID NO:2.

78. The method of claim 76, wherein the C2GnT polypeptide has the amino acid sequence of SEQ ID NO:2.

79. The method of claim 76, wherein said compound comprises a nucleotide-sugar, a dolichol-phosphate-sugar, or a dolichol-pyrophosphate-oligosaccharide moiety.

80. The method of claim 76, wherein the acceptor is selected from the group consisting of a saccharide, an oligosaccharide, a polysaccharide, a glycopeptide, a glycopolypeptide, and a glycolipid.